

DNA and CHROMOSOMES

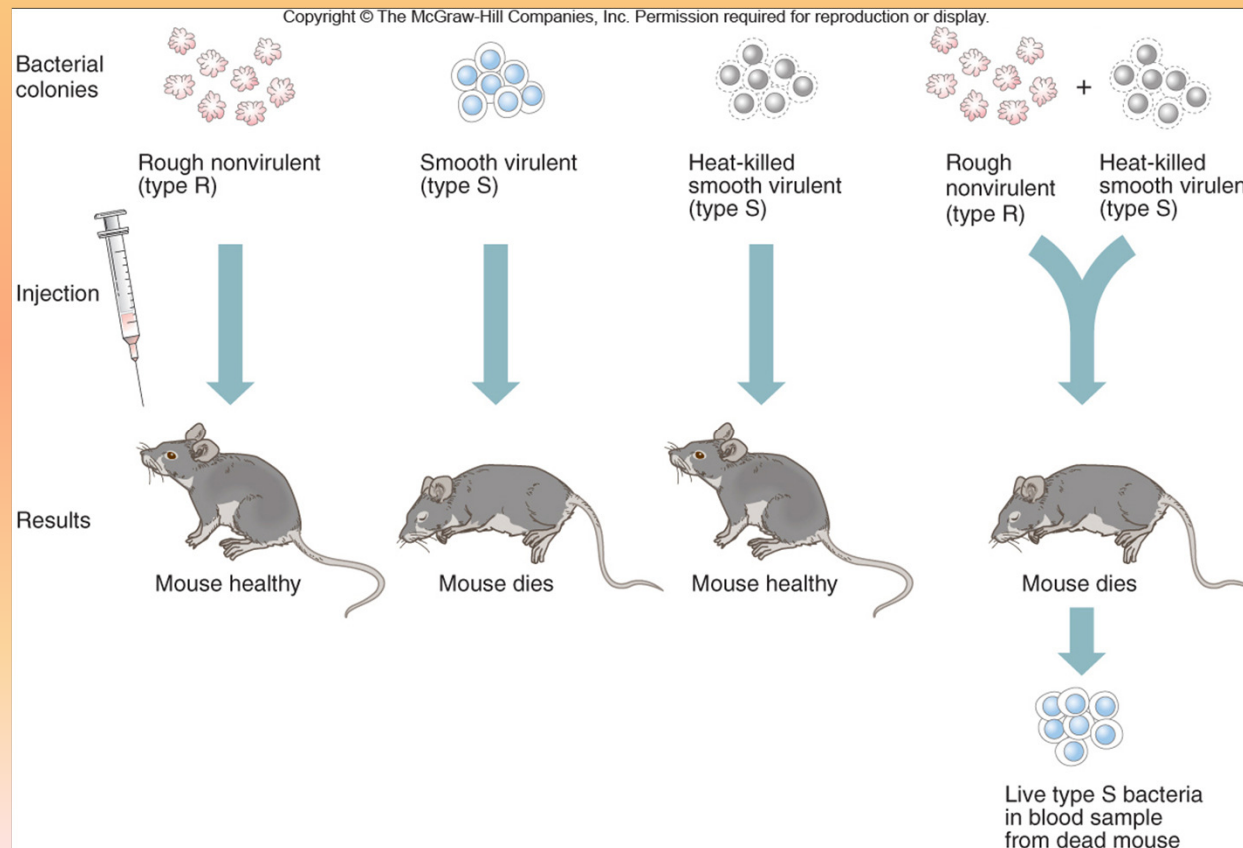
Chapter 12

Identifying the Substance of Genes

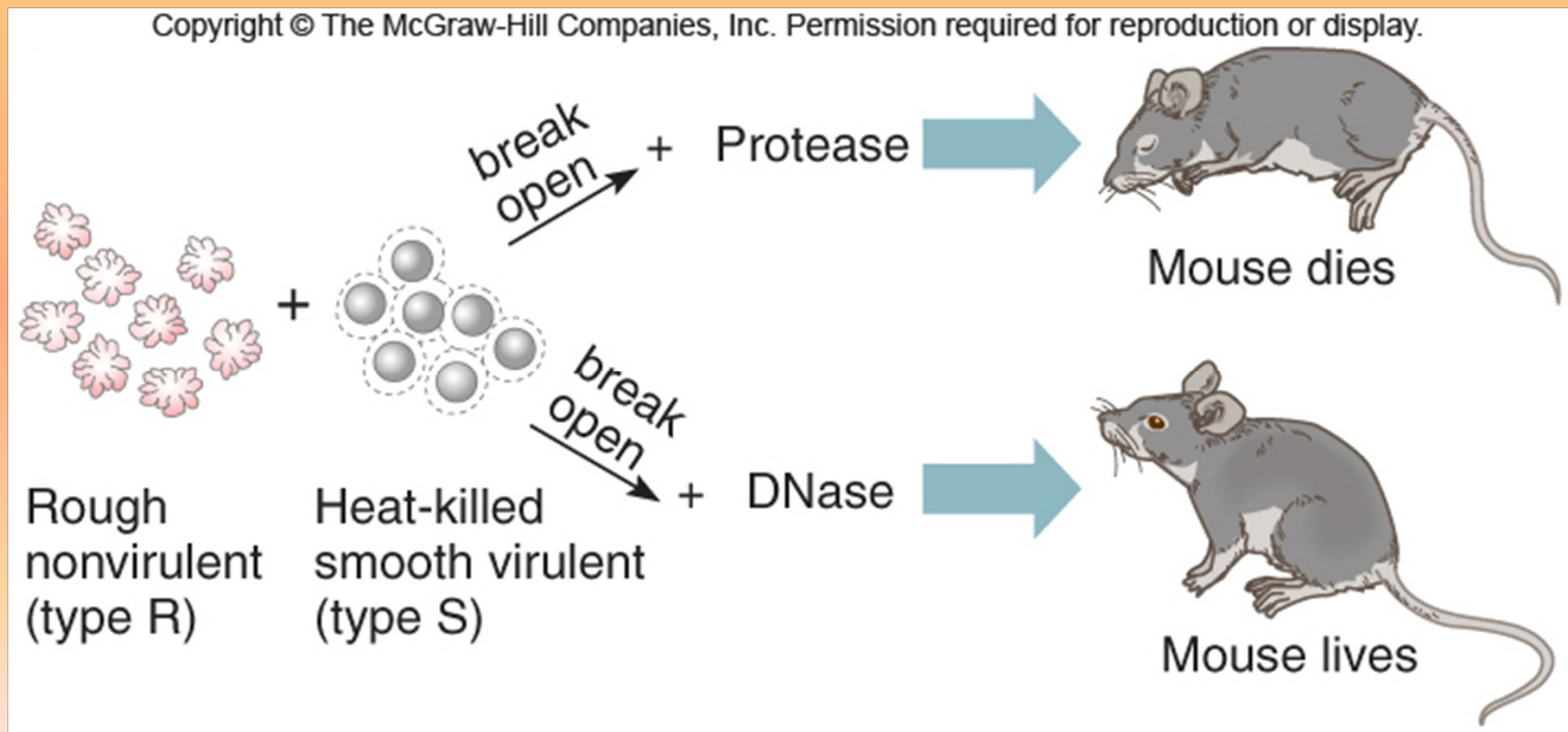
- Griffith

- **Transformation** =

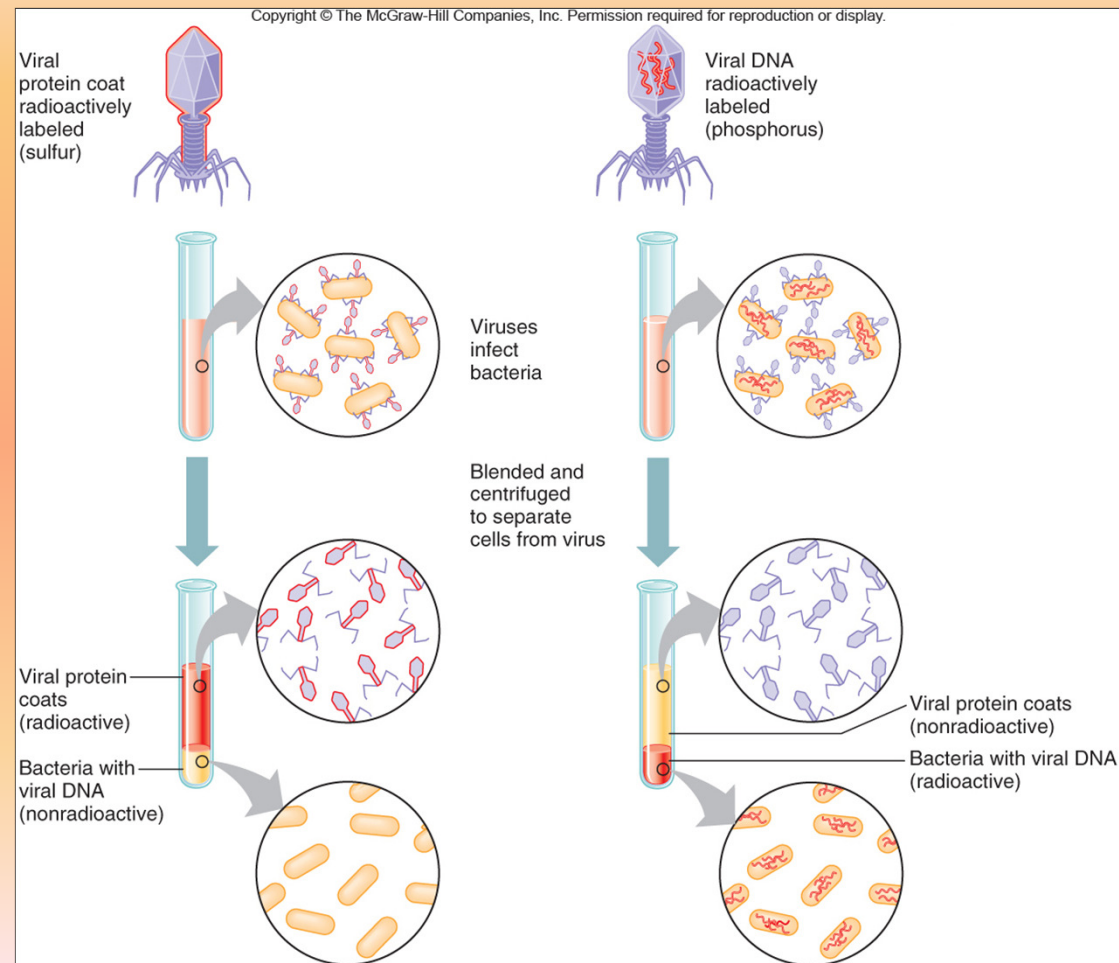
- One strain of bacteria was changed by genes from another strain of bacteria



- Avery, MacLeod, McCarty
 - Determined DNA stores and transmits genetic info from one generation of bacteria to the next



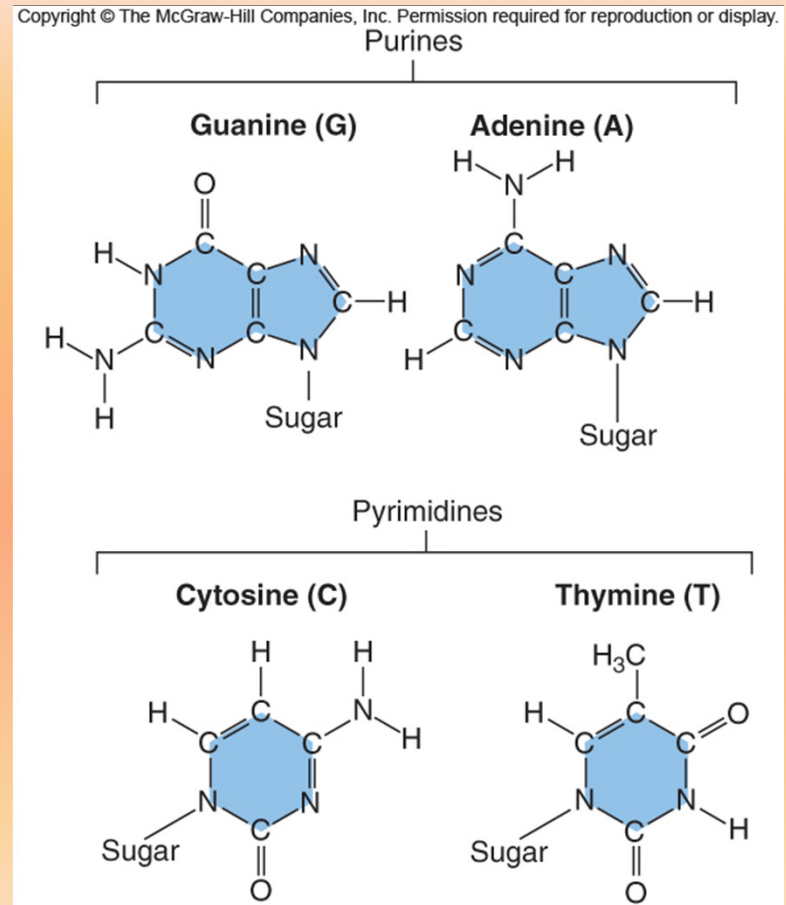
- Hershey and Chase
 - Confirmed DNA (not protein) is the genetic material using radioactive isotopes in viruses



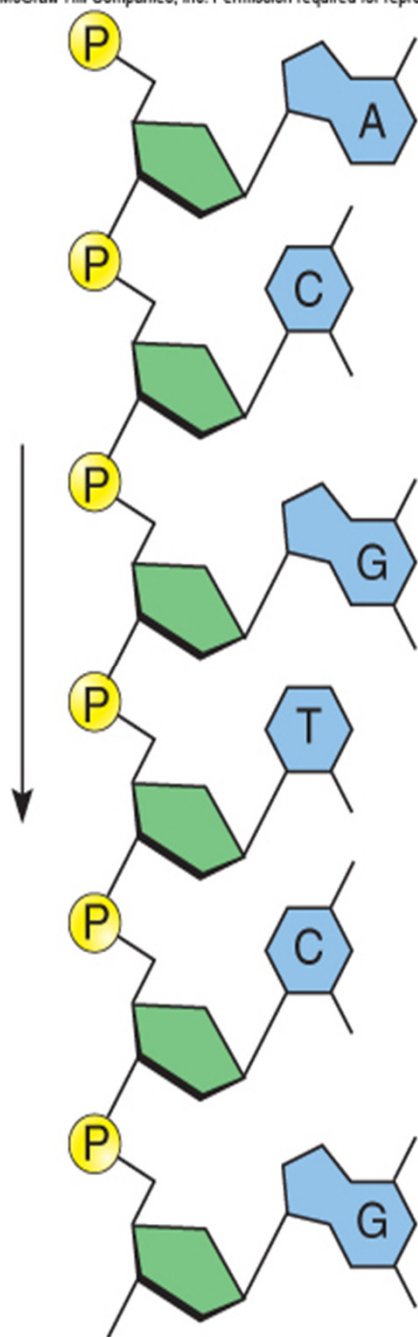
- Conclusions
 - DNA makes up genes
- Assumptions
 - DNA must be capable of
 - Storing information
 - Copying information
 - Transmitting information

The Structure of DNA

- Building blocks of nucleic acids = **nucleotides**
 - 1 deoxyribose sugar
 - 1 phosphate group
 - 1 nitrogenous base
 - Adenine (A)
 - Guanine (G)
 - Cytosine (C)
 - Thymine (T)

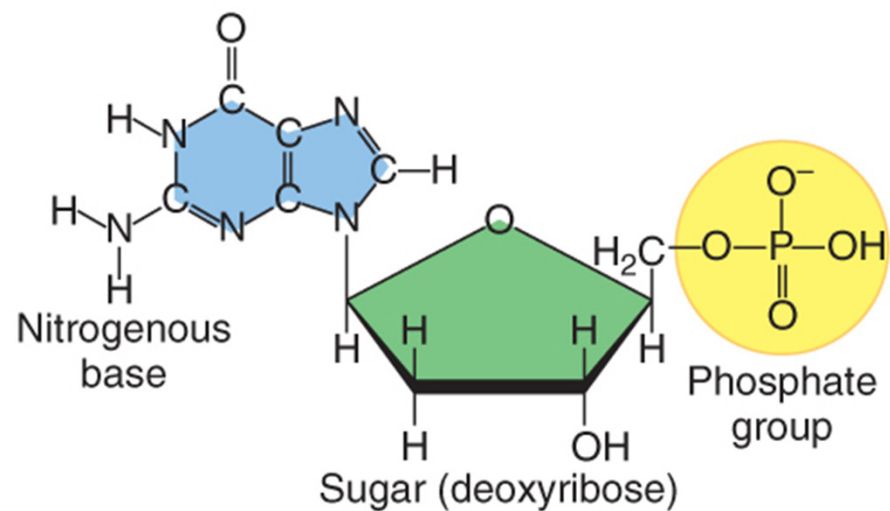


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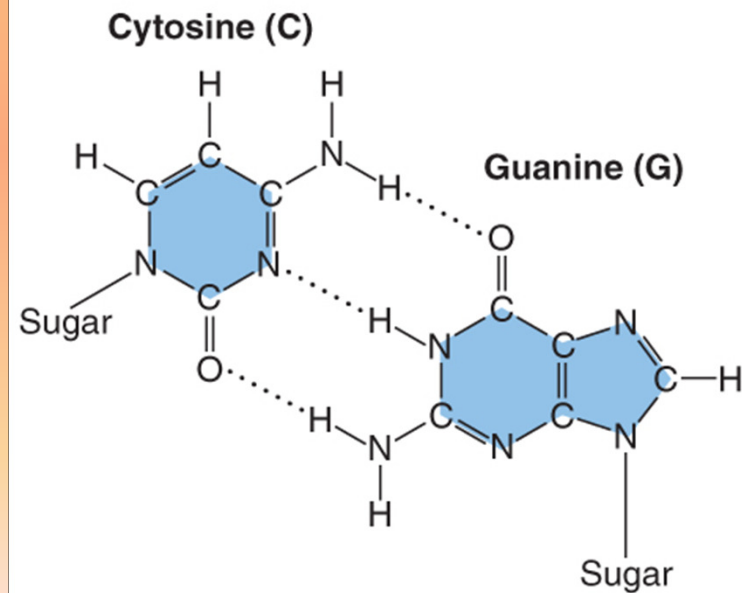
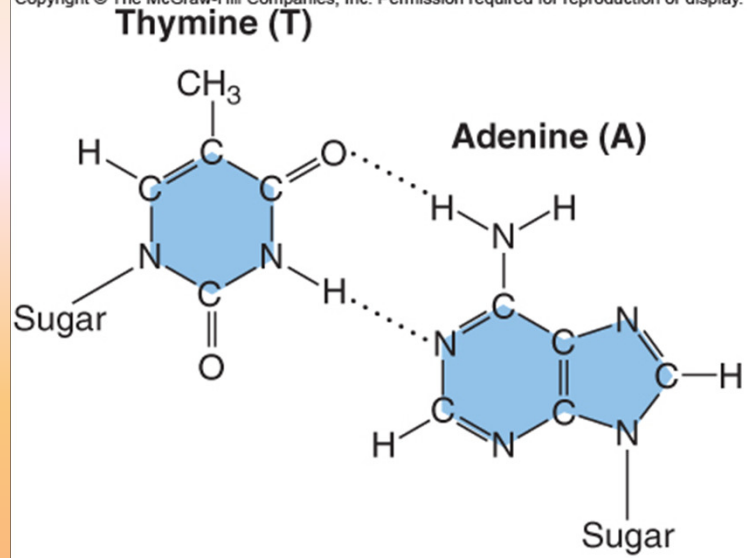
Guanine (G)



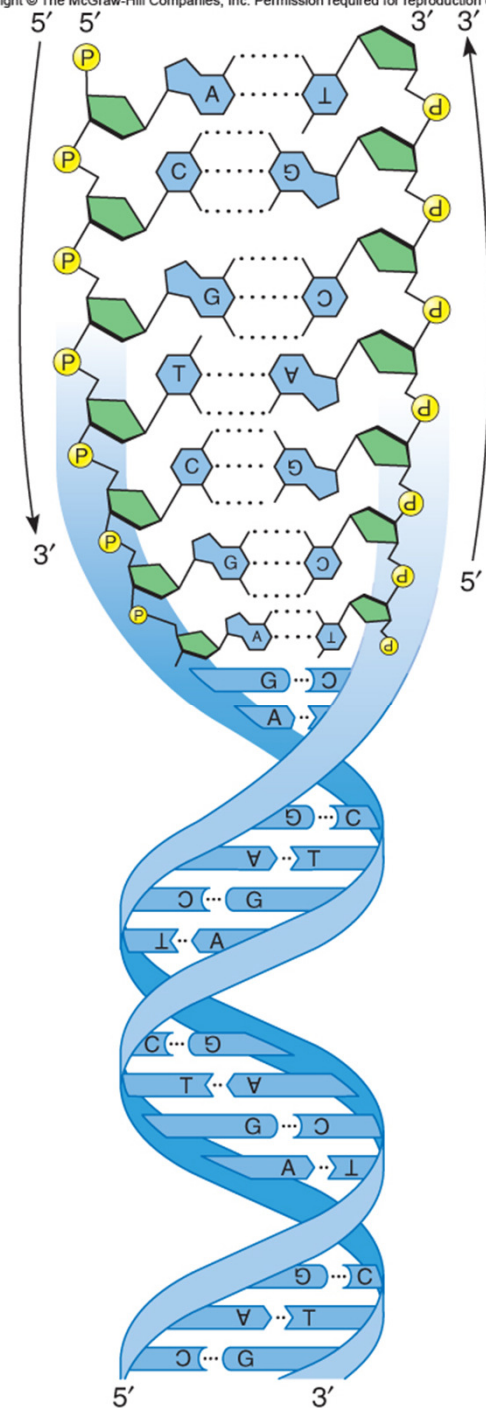
- Chargaff's Rule
 - $A = T$
 - $C = G$
- Wilkins and Franklin
 - Work with X ray diffraction showed patterns indicating DNA's shape is a helix
- Watson and Crick
 - Determined 3-D structure of DNA is a double helix

- Nucleotides join to form long chains
 - Sugar-phosphate backbone
 - Strands are antiparallel
 - 5 prime (5') and 3 prime (3') ends
 - Nitrogenous bases held to those of 2nd strand by hydrogen bonds
 - Complementary base pairs
 - **A bonds with T**
 - **C bonds with G**
- Double helix forms when antiparallel, base-paired strands twist

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- **Chromatin** =

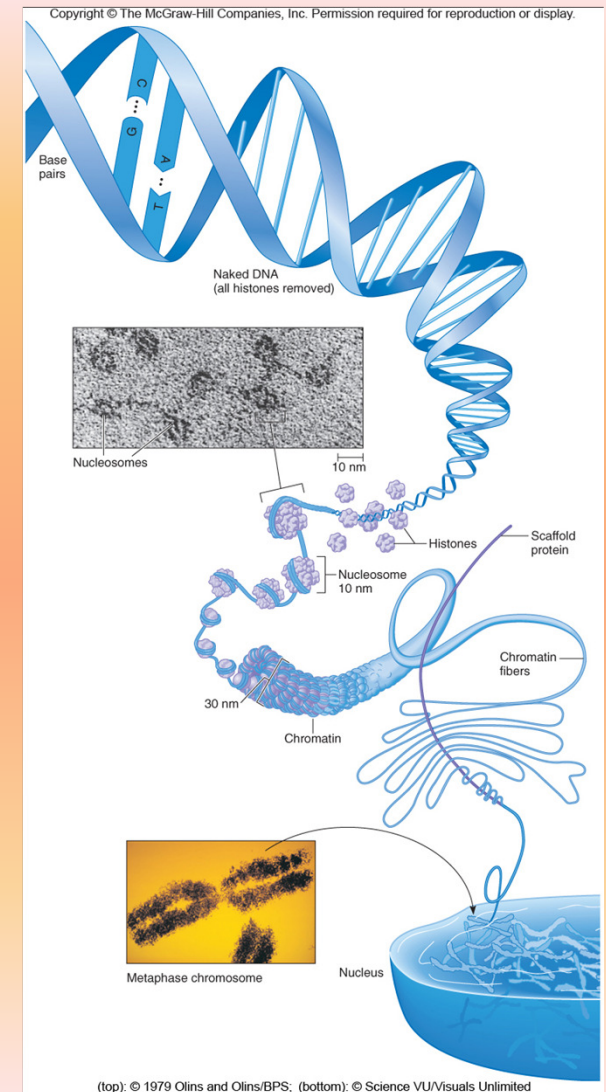
- Complex consisting of DNA tightly coiled around proteins called **histones**

- **Nucleosome** =

- Bead-like unit of chromatin structure

- DNA wraps at several levels, compacting into a chromosome

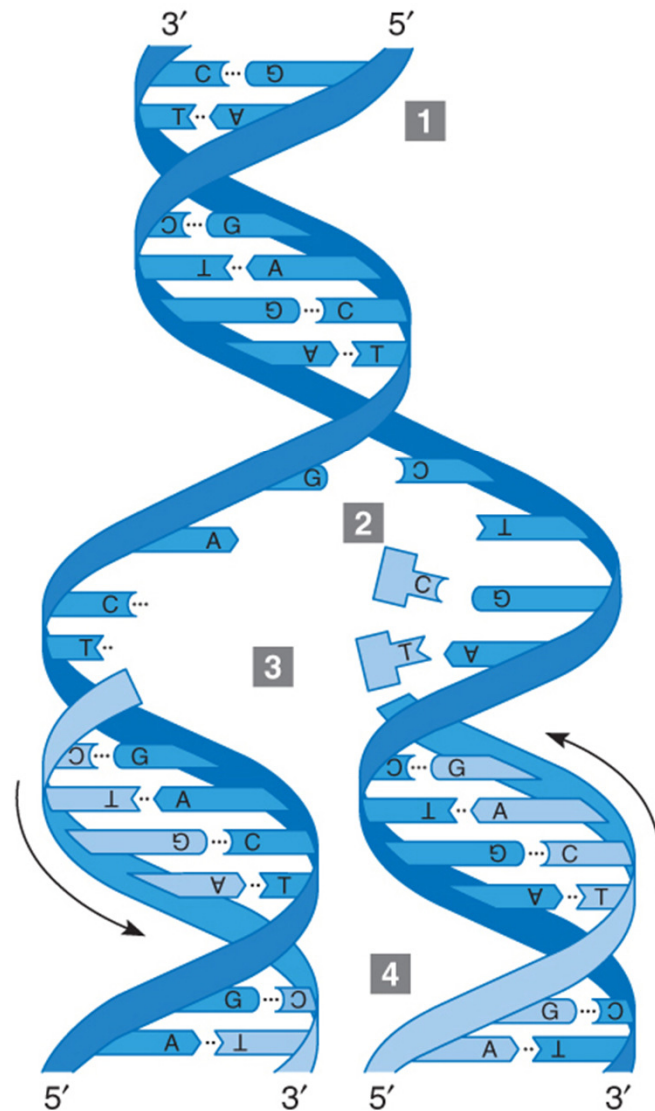
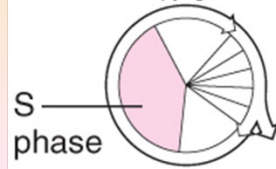
- Very tightly packed during cell division



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DNA Replication

- **Semiconservative replication** =
 - Each replicated DNA molecule has one original strand and one new strand
- Overview
 1. Strands unwind and separate at several points
 2. **DNA polymerase** adds complementary bases to template
 - **A with T and vice versa**
 - **G with C and vice versa**
 3. Sugar-phosphate backbones seal back up
- Result
 - 2 DNA molecules
 - Identical to each other and original



1 Parent DNA molecule.

2 Parental strands unwind and separate at several points.

3 Each parental strand provides a template for DNA polymerase to bind complementary bases, A with T and G with C.

4 Sugar-phosphate backbones of daughter strands close.

- DNA Replication occurs during the S phase of the cell cycle
- Prokaryotes
 - Replication starts from a single point and proceeds in two directions until finished
- Eukaryotes
 - Replication occurs simultaneously from multiple starting points in both directions

- **Replication fork** =

- Locally opened portions of DNA double helix

- DNA polymerase works directionally

- Only adds new nucleotides to exposed 3' end

- Replication proceeds in a 5' to 3' direction

- Replication on one strand is continuous

- The other is discontinuous

- Produces small pieces called **Okazaki fragments**



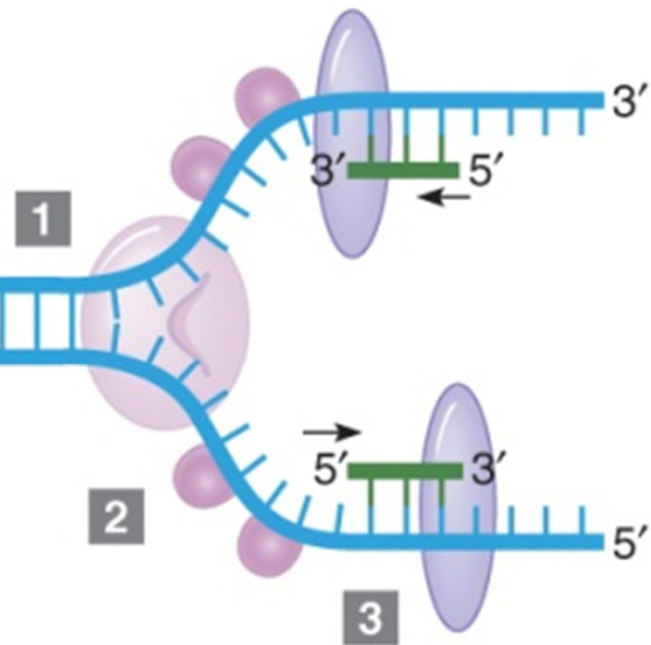
Activities at the Replication Fork

1 Helicase binds to origin and separates strands.

5'
3'

2 Binding proteins keep strands apart.

3 Primase makes a short stretch of RNA on the DNA template.



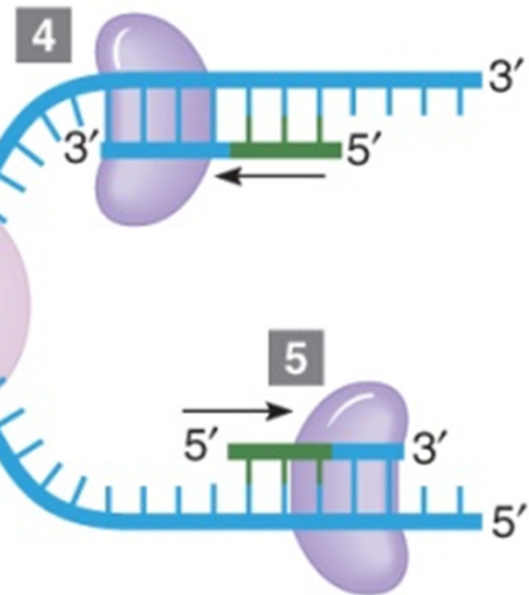
Activities at the Replication Fork

4 DNA polymerase adds DNA nucleotides to the RNA primer.

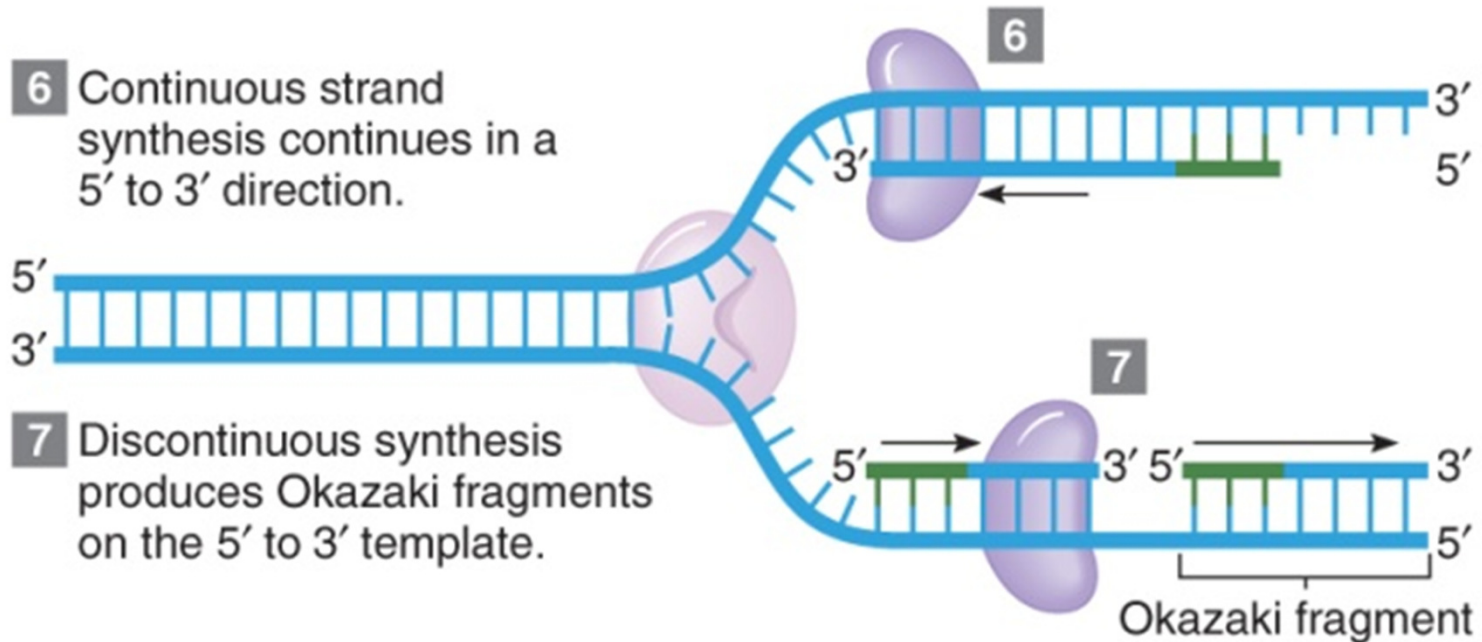
Overall direction of replication

5'
3'

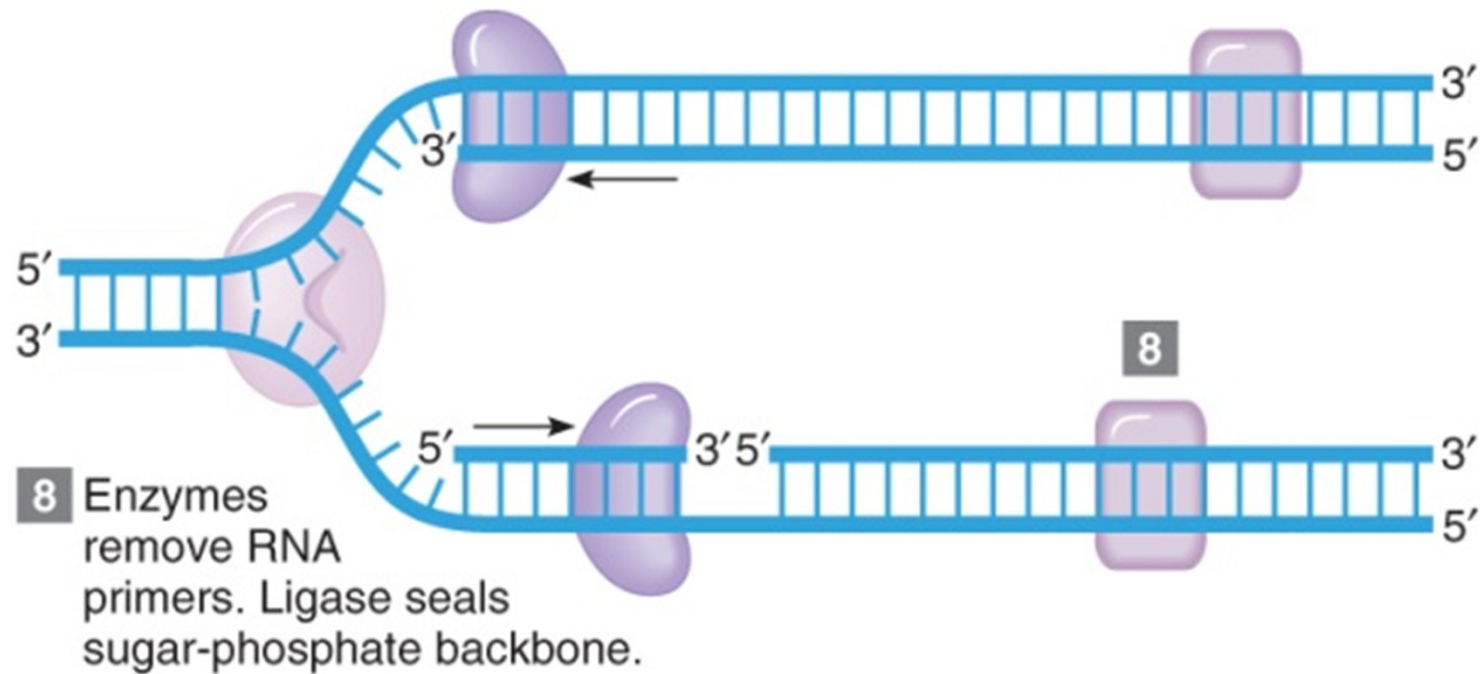
5 DNA polymerase proofreading activity checks and replaces incorrect bases.



Activities at the Replication Fork

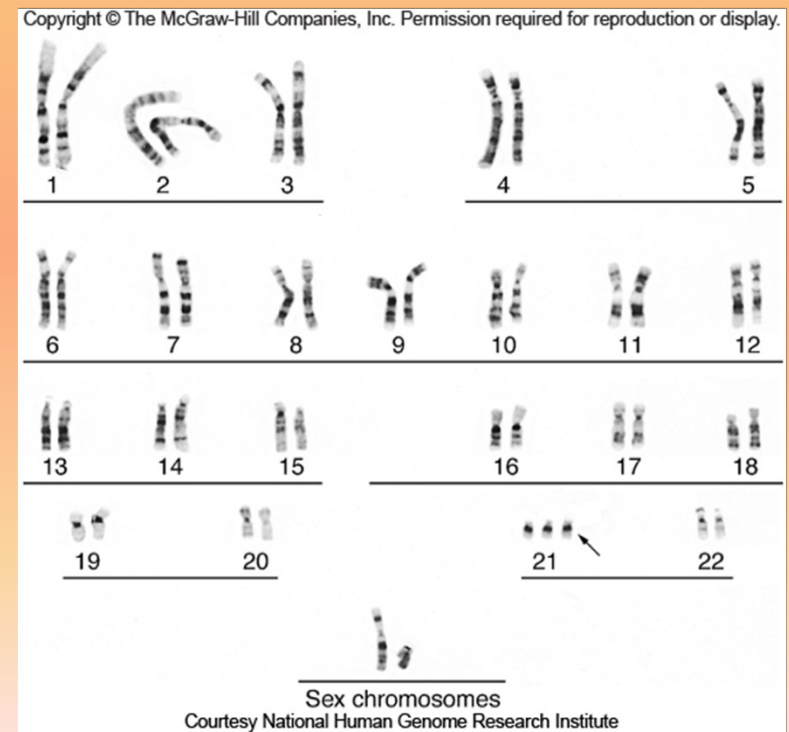


Activities at the Replication Fork



Chromosomes

- **Genome** =
 - Entire set of genetic information that an organism carries in its DNA
- **Karyotype** =
 - Shows complete diploid set of chromosomes
 - Grouped in pairs in order of decreasing size
 - Humans have 46 total chromosomes
 - 2 sex chromosomes
 - XX = female
 - XY = male
 - 22 pairs of autosomes



Chromosomal Disorders

Abnormal Chromosomal Number

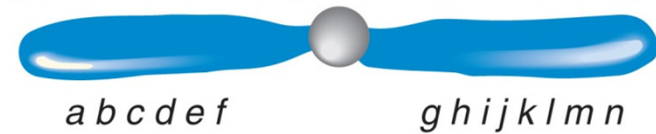
- **Nondisjunction** =
 - Homologous chromosomes do not separate properly
 - Error in anaphase of meiosis
 - Results in abnormal chromosome number
- **Monosomy** =
 - One missing chromosome
- **Trisomy** =
 - One extra chromosome

- Nondisjunction of autosomes could result in:
 - Down syndrome (Trisomy 21)
 - Mental retardation and birth defects
 - Edward syndrome (Trisomy 18)
 - Patau syndrome (Trisomy 13)
- Nondisjunction of sex chromosomes could result in:
 - Turner syndrome (XO)
 - Sterile female
 - Klinefelter syndrome (XXY)
 - Male (typically unable to reproduce)
 - Note: Y with no X won't even be born

Abnormal Chromosome Composition

- **Deletion** =
 - Loss of part of a chromosome
- **Duplication** =
 - Extra copy of a part of chromosome
- **Inversion** =
 - Reverses direction of parts of chromosome

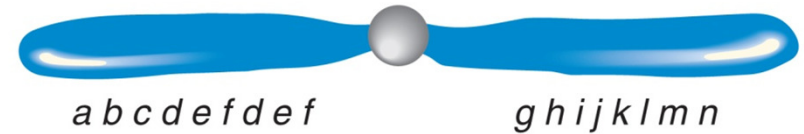
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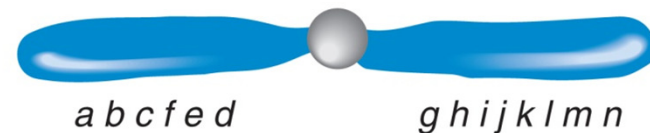
a. Normal sequence of genes



b. Deleted sequence of genes



c. Duplicated sequence of genes



d. Inverted sequence of genes

- **Translocation** =
 - Part of one non-homologous chromosome breaks off and attaches to another
- **Insertion** =
 - Insertion of a larger sequence into a chromosome
 - Due to unequal crossing over during meiosis